

Opis choroby *

Definicja

A progressive cutaneous disorder predominantly affecting males, characterized by hypertrophy and thickening of the skin of the scalp, forming convoluted furrows with deep, tender, and cerebriform cutaneous folds. Hair is usually normal in the furrows and sparse on the folds. It can be isolated or associated with other abnormalities, such as intellectual deficit, epilepsy, cataract, blindness, and deafness.

Dane

Klasyfikacja

Grupa fenomenów

Kod ORPHA

671

Kod OMIM

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Kod ICD10

Q82.8

Kod ICD11

EE7Y

*Źródło

orphanet